

SCREENING OF PROTOONCOGENE RET IN MOROCCAN PATIENTS WITH MEDULLARY THYROID CARCINOMA

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Background/Purpose: To investigate the contribution of proto-oncogene *RET* gene to medullary thyroid carcinoma in Moroccan families.

Methods: A total of 31 probands with MTC, from 31 unrelated families and 113 of their relatives were included in this study. Seven (22.5%) probands were with multiple endocrine neoplasia type 2A (MEN2A), three (9.7%) with MEN2B and 11 (35.4%) with isolated MTC. The majority of index cases were referred to the endocrine department for a post-operative management. The exons 5, 8, 10, 11 and 13–16 of *RET* were directly sequenced.

Results: A total of seven distinct heterozygote missense mutations were identified in *RET* exons 11, 14, 15 and 16. All of these mutations were previously described as disease-causing in different populations. These mutations were identified in 100% of patients with MEN2A, 100% of patients with MEN2B and 36% of isolated MTC (27% were familial cases). The most affected codon was 634 (50%), followed by codons 918 (22%), 804 (14%) and 891 (14%). Among at risk subjects, 18.5% were as mutation carriers. Fourteen patients including seven children were asymptomatic. Four of them were submitted to prophylactic thyroidectomy.

Discussion & Conclusion: This is the first comprehensive genetic analysis of MTC among Moroccan families. Our results will contribute to the definition of a national policy of this cancer control by encouraging earlier diagnosis and management of patients with MTC in Morocco.